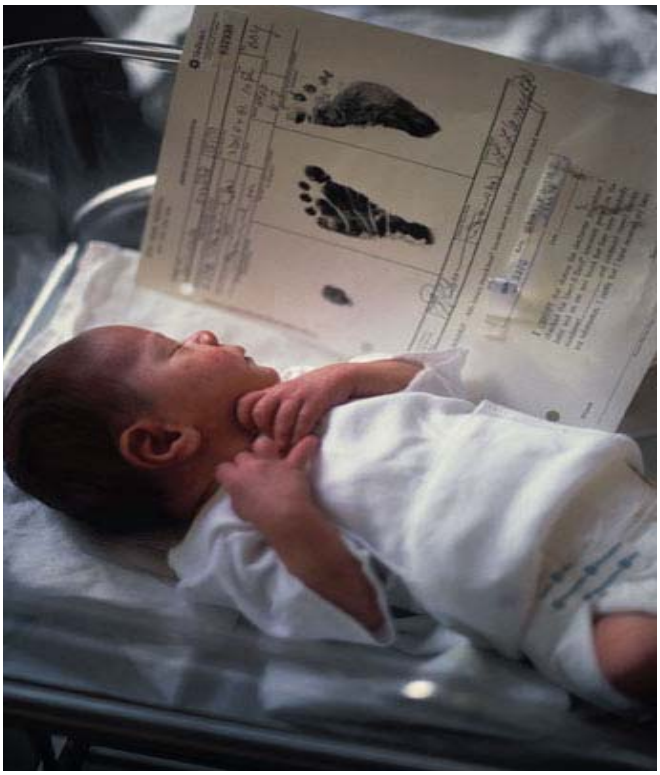

PARENT'S GUIDE TO YOUR BABY'S NEWBORN SCREENING

NEBRASKA NEWBORN SCREENING PROGRAM

(Updated Jan 2006)

NEBRASKA HEALTH AND HUMAN SERVICES SYSTEM

WHAT IS NEWBORN SCREENING?

Newborn screening is routinely done for every baby born in Nebraska. It is a set of blood tests done to find certain disorders. You can't tell by looking at your baby if they have any of these disorders. The blood tests are needed to find them. Newborn screening has been done in the United States and around the world since the 1960's, but more disorders are screened for today.

Who needs to be screened? All babies born in Nebraska must have the blood test to see if they have certain disorders.

How is my baby screened? A small sample of blood is taken from your baby's heel. The blood sample is used to test for the disorders described in this booklet.

Why are the screening tests important?
The law was written to help prevent mental retardation and other damaging problems in children. The disorders screened for can usually be controlled by proper treatment. Treatment can help prevent mental retardation, illness or even death. *(There are other causes of mental retardation, illness and death which will not be found by these tests).*

When should my baby be screened? Your doctor will have the blood collected for the tests before your baby leaves the hospital. If you and your baby go home before your baby is 24 hours old, your baby will need to have some tests repeated within one week. The hospital may schedule this appointment before you leave. If not, you should contact your doctor to have some of the tests repeated within one week.

Babies born at home: Babies who are born at home must also be screened for these diseases. The parents or the person registering the birth must arrange with a doctor to have the tests completed. The test should be done between 24 and 48 hours of birth, so that any needed treatment can be started as soon as possible. If a baby is older they should still get the screening. Even though it is less ideal in terms of timing, older babies may still benefit from treatment for certain disorders.

WHAT DISORDERS ARE INCLUDED IN NEBRASKA'S ROUTINE SCREENING PANEL?

Biotinidase Deficiency

(BYE-o-TIN-I-days) is a disorder found in babies who are missing the enzyme biotinidase. Babies who are missing the enzyme need more biotin than is normally found in the diet. This disorder can lead to seizures, developmental delay, eczema, and hearing loss. Problems can be prevented with biotin treatment started in the first few weeks of life.

Congenital Adrenal Hyperplasia

(Congenital A-DREE-nal Hyper-PLAY-zia) is a disorder caused by an enzyme deficiency that results in the adrenal gland producing too little aldosterone and cortisol and too much androgen. Babies with the Salt-wasting form of this disorder are at risk of an adrenal crisis that can cause sudden death. Problems can be prevented with hormone treatment started early.

Congenital Primary Hypothyroidism

(HIGH-po-THIGH-roid-ism), or CPH is a disorder caused by not having enough thyroid hormone. Babies with CPH often appear normal at birth. The most common effects of CPH are mental retardation and the child not growing normally. If treatment with thyroid medication starts in the first few weeks of life, these children usually develop normally.

Cystic Fibrosis

(SIS-tic FY-bro-sis) is a genetic disorder that affects people in a variety of ways. They may have persistent coughing, wheezing or shortness of breath, an excessive appetite but poor weight gain and greasy, bulky stools. If they have a problem with the pancreas affecting weight gain, early treatment can improve the child's growth and development. With early diagnosis from newborn screening, some may have fewer hospitalizations, and regular monitoring may prevent or reduce lung infections.

Galactosemia

(Guh-LAK-toe-SEE-me-ah) is a disorder in which a simple sugar called "galactose" can't be broken down in the body. Galactose is found in breast milk, many formulas and milk products. If it remains at high levels in the body and is not broken down, it can harm the baby's eyes, liver, and brain. In some cases life threatening damage to the brain and liver can occur. When started early, a special diet can prevent these problems.

Hemoglobinopathies

(He-ma-glow-bin-OP-ah-thees) are a group of disorders of the red blood cells including sickle cell anemia. Babies with this are more likely to have anemia, episodes of pain, strokes, and life-threatening infections. Early treatment with antibiotics, immunizations and parent

education can help. These measures can prevent serious infections in childhood, and improve outcomes for babies with this disorder.

Medium Chain Acyl Co-A Dehydrogenase Deficiency (MCAD)

(Medium Chain A-seal Co-A Dee-HIGH-dra-gen-AZE Deficiency - MCAD) is a disorder of fatty acid metabolism. When babies and children with MCAD become ill or have long periods of fasting, blood sugars become dangerously low and they are at risk of having a "metabolic crisis." A metabolic crisis can lead to seizures, failure to breath, cardiac arrest and death, and/or result in serious brain damage. However, screening can provide diagnosis before symptoms occur. Parents can then prevent the fasting periods, and know when to seek early medical care, to prevent the crisis. A special diet supplement is often added to the baby's diet to help prevent problems.

Phenylketonuria (PKU)

(FEE-nil-KEE-tone-u-ree-ah), or PKU, is a disorder caused when the body can't break down phenylalanine. Phenylalanine, or phe, is an amino acid found in protein foods like milk, meats, eggs and cheese. In babies with PKU, the phe remains at high levels in the body after eating these foods. These high levels may cause nerve and brain cell damage. This damage can result in mental retardation. If detected early and the baby is started on a special low phenylalanine diet, mental retardation is prevented.

What if any of the screening results are abnormal?

A "positive" or abnormal screening test result only means that your baby *might* have one of the disorders mentioned above. A diagnosis of a disorder is usually *not* made with the first lab test. Further testing will be necessary to determine if your baby actually has the disorder.

If you are asked to have your child re-tested, please act quickly so tests can be repeated and final results obtained. If needed, treatment must be started as soon as possible to prevent the onset of mental retardation or other damaging results.

A positive (abnormal) screening result means your baby should have more testing, or be retested. It does NOT mean your baby has the disorder. Sometimes positive screening results are found in babies that do not have the disorder. This is called a false positive screening result.

Why else might I be asked to have my baby "re-tested?"

Some things can cause problems with the tests. When these things happen, we can't be very sure of some of the results. The most common reasons why parents are asked to bring their babies back for re-testing are:

1) The specimen was collected too early (less that 24 hours of age).
2) The specimen wasn't collected before a blood transfusion; or
3) A problem occurred with the quality of the specimen (e.g. problem with collection and/or handling of the dried blood spots).

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"SUPPLEMENTARY" SCREENING

What is "supplementary" screening?

➢ The supplementary screening or "Tandem Mass Spectrometry" testing can produce results on about 30 disorders. For *most* of these, intervention or medical treatment can help prevent the complications or damage from the disorder. However, for *a few* of these disorders, because they are so rare what we know about them and about the best medical treatment is limited.

➢ The State of Nebraska cares about the health of your child, so the newborn screening program has arranged with the laboratory so that you can get these screening results with no additional blood and for no additional cost.

➢ The March of Dimes and the American College of Medical Genetics recommend screening newborns and/or reporting results for all of these disorders that can be found with the Tandem Mass Spectrometry test. The disorders screened for on the supplementary panel are:

<table>
<tr>
<td>Organic Acid Disorders:
❖ 3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency (HMG)
❖ Glutaric Acidemia-Type I (GA-I)
❖ Isobutyrl-CoA Dehydrogenase Deficiency
❖ Isovaleric Acidemia (IVA)
❖ 2-Methylbutyryl-CoA Dehydrogenase Deficiency
❖ 3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC Deficiency)
❖ 3-Methylglutaconyl-CoA Hydratase Deficiency
❖ Methylmalonic Acidemias
❖ Mitochondrial Acetoacetyl-CoA Thiolase Deficiency (3-Ketothiolase Def.)
❖ Propionic Acidemia (PA)
❖ Multiple CoA Carboxylase Deficiency
❖ Malonic Acidemia</td>
<td>Fatty Acid Oxidation Disorders:
❖ Carnitine/Acylcarnitine Translocase Deficiency (Translocase)
❖ 3-Hydroxy Long Chain Acyl-CoA Dehydrogenase Deficiency (LCHAD)
❖ Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD) (required test)
❖ Multiple Acyl-CoA dehydrogenase deficiency (MADD or GA II)
❖ Neonatal Carnitine Palmitoyl Transferase Deficiency Type II (CPT-II)
❖ Short Chain Acyl-CoA dehydrogenase deficiency (SCAD)
❖ Short Chain Hydroxy Acyl-CoA Dehydrogenase Deficiency (SCHAD)
❖ Trifunctional protein deficiency (TFP deficiency)
❖ Very long-chain Acyl-CoA dehydrogenase (VLCAD)</td>
</tr>
<tr>
<td>Amino Acid Disorders:
❖ Argininosuccinate Aciduria (ASA Lyase deficiency)
❖ Citrullinemia (ASA Synthetase deficiency)
❖ Homocystinuria
❖ Hypermethioninemia
❖ Maple syrup urine disease (MSUD)
❖ PKU (required test)
❖ Tyrosinemia</td>
<td>Other Abnormal Profiles:
❖ Hyperalimentation
❖ Liver Disease
❖ Medium Chain Triglyceride (MCT) Oil Administration
❖ Presence of EDTA Anticoagulants in blood specimen
❖ Treatment with Benzoate, Pyvalic Acid, or Valproic Acid
Note: The screening laboratory specifies that non-ketotic hyperglycinemia (NKH) cannot be reliably detected by tandem mass spectrometry technology.</td>
</tr>
</table>

CONSENT FOR SUPPLEMENTARY SCREENING

You will have to decide if you wish to consent to the supplemental screening test or not. If you do consent, the laboratory test results will be shared with your baby's physician, and if anything unusual should appear, additional tests may be recommended. If you decide you do not want to have your baby tested for the other *supplemental* disorders, you will be asked to sign a dissent form indicating your wishes. If you dissent from supplemental screening, your baby will only be tested for the 8 required disorders.

➢Note: The instrument used to screen for MCAD and PKU has several "markers" that it looks at. This is done to have the best chance of identifying MCAD and PKU. Some of these markers may indicate an abnormality for something other than MCAD or PKU. Repeat testing will be recommended in these cases.

* *

PROTECTIONS FOR YOUR BABY'S BLOOD SPECIMEN

It is important that you feel confident your baby's blood specimen is used only for its intended purpose. That is, to find information that can help your baby. Currently laboratories must keep the newborn screening specimens at least 90 days. After this period the laboratory has 30 days to dispose of the sample. The blood spots are disposed of in a way that they can not be linked to identifying information. This is an important protection of genetic information.

Your baby's blood specimen can not be used for medical research without your written consent. The laboratory can only release a newborn specimen for medical research if your baby's confidentiality is preserved. Medical research using any newborn screening specimen must follow protections of human subjects from research risks under subpart A of part 46 of 45 Code of Federal Regulations, as they existed on September 1, 2001.

If you give consent for the "supplemental" screening, this does NOT give permission for use of the specimen for research. A separate consent must be used for any request to use your newborn's specimen for research.

* *

The newborn screening tests are not diagnostic. They are a "screen", designed to detect newborns who need further testing to determine if they have certain disorders. The screening tests are very efficient and provide newborns with the best opportunity for having the disorders identified early. However, like most laboratory tests, the tests used for newborn screening can not guarantee that every affected newborn will be identified, or that only infants at higher risk of being affected will be identified. Therefore, it is important to recognize that there will be some "false positives" (newborn with a positive or abnormal screen result who are later found to have normal results), and the possibility of "false negatives" (affected newborns whose screening test results do not indicate an underlying disorder).

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NEWBORN HEARING SCREENING

All hospitals offer newborn hearing screening during the birth admission or by arrangement with another facility after the birth admission.

Significant hearing loss is one of the most common major birth defects. If not found early, hearing loss will delay speech, language and cognitive development in children. Early detection of hearing loss along with early intervention and treatment before six (6) months of age is highly effective in helping a child's language, communication and educational development.

The hearing screening test is a safe procedure that can be done in a very short time. There are two methods that might be used: auditory brainstem response (ABR) and/or otoacoustic emissions (OAE). Both are done when the baby is asleep or quiet. Responses to sounds sent through tiny earphones are counted and averaged by a computer. It is important to remember, that while many newborns may not pass the hearing screening test, only further audiological testing and evaluation can determine which ones really have hearing loss. So it is important that you work with your baby's physician to get further testing if the screening results indicate "refer" (or did not pass).

If your baby passes the hearing screen, but has risk factors you and your baby's physician will want to monitor your child closely. Some risk factors include: a family history of hearing problems, low birth weight or certain other medical conditions. The newborn hearing screening test will not pick up hearing loss that develops later, (for example as a result of serious infection or illness).

If your child is suspected of or found to have a hearing loss, Nebraska's Early Intervention Program called the "Early Development Network" can help you coordinate services with local school systems and providers for evaluations, early intervention services and assistive technology.

For More Information on HEARING SCREENING:

Refer to the brochure: "The Nebraska Newborn Hearing Screening Program", included with your newborn packet. If you did not receive this brochure call the NNHSP phone number below:

For questions about Nebraska's **Newborn Hearing** Screening Program (NNHSP), contact the Nebraska Department of Health and Human Services at : 1-402- 471-6733.

For questions about Nebraska's **Early Intervention** Program contact "Nebraska CHILDFIND" at 1-888-806-6287. Or go to www.nde.state.ne.us/ECH/EARLY/echp.htm.

For More Information on NEWBORN (Blood-spot) SCREENING

For questions about Nebraska's Newborn Screening Program call 1-402-471-6733 or 1-402-471-0374 or e-mail newborn.screening@hhss.ne.gov. Also, check out our web site at: www.hhss.ne.gov/nsp/

To order more PARENT INFORMATION booklet/brochures:

Call 402-471-9731 or fax order to "NNSP" : fax 402-471-1863 or e-mail newborn.screening@hhss.ne.gov. **Specify: Parent's Guide booklet, or Hearing Screening Brochure. Also available in several other languages.**

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